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First trimester screening versus non-invasive prenatal testing (NIPT)

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Several tests with multiple components are being used for the antenatal screening of fetal aneuploidy. Triple test (second trimester screening) screening started in 1990 using three biochemical markers together with the maternal age for the risk calculation. A significant improvement in the risk assessment has been achieved by introducing the first trimester screening (FTS) in the year 2000. The algorithm uses mainly the maternal age, the nuchal translucency (NT) together with 2 biochemical markers, free beta-HCG and PAPP-A.

NIPT testing based on the detection of cell free fetal DNA in maternal blood has improved significantly over the past 3 years. The currently used testsystems detect trisomies 21, 18 and 13 as well as X and Y. The specificity is reported to be >99%. The sensitivity to detect T21 is >99%, T18 > 97% and T13 >80%. The sensitivity for detecting the female gender is >90%, the male gender is recognized with a sensitivity of >99%.

NIPT is especially indicated if the triple test or the first trimester screening indicate an increased risk for T21 or T18, at increased maternal age and in cases of anxiety towards invasive procedures (AC / CVS).

NIPT is not the method of choice if there are specific fetal anomalies on ultrasound, in triplet pregnancies, in vanished twin and in cases of known genetic anomalies that cannot be diagnosed by NIPT.